

Health Care Provider Fact Sheet

Disease Name

Galactosemia

Acronym

GALT

Disease Classification

Disorder of carbohydrate metabolism

Variants

Yes

Variant name

Duarte galactosemia

Symptom onset

Infancy

Symptoms

The affected infant may appear normal at birth. Within a few days to two weeks after initiating milk feedings, the infant develops vomiting, diarrhea, lethargy, jaundice, and liver damage. Untreated, the disorder may result in death, frequently associated with *E. coli* septicemia. Infants surviving the above symptoms may evidence developmental retardation, hepatomegaly, Fanconi's syndrome, growth failure and cataracts.

Natural history without treatment

If not detected immediately, it results in liver disease, cataracts, mental retardation, and even death. Death can occur as early as one to two weeks of age from severe escherichia (*E. coli*) bacteria infections. *E. coli* infections are common in untreated galactosemic infants. The American Liver Foundation recommends that all infants who develop jaundice be considered for galactosemia.

Natural history with treatment

As Galactosemic children get older they may encounter delays in speech and females may suffer from ovarian failure. Nevertheless, children who are diagnosed early have very good long-term outlooks and will lead normal, healthy lives.

Treatment

Treatment for galactosemia is the elimination of galactose and lactose from the diet throughout life. Infants are placed on soy formula.

Emergency Medical Treatment

See sheet from American College of Medical Genetics (attached) or for more information, go to website:
<http://www.acmg.net/StaticContent/ACT/Galactose+GALT.pdf>

Physical phenotype

No abnormalities present at birth.

Inheritance

Autosomal recessive

General population incidence

1:65,000 live births

OMIM Link

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=606999>

Genetests Link

www.geneclinics.org

Support Group

Parents of Galactosemic Children, Inc.
<http://www.galactosemia.org>

Children's Liver Alliance
<http://www.liverkids.org.au>

Children Living with Inherited Metabolic Diseases
<http://www.climb.org.uk/>

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